



Case Report

Structural Characteristics in the γ Chain Variants Associated with Fibrinogen Storage Disease Suggest the Underlying Pathogenic Mechanism

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Abstract: Particular fibrinogen γ chain mutations occurring in the γ -module induce changes that hamper γ - γ dimerization and provoke intracellular aggregation of the mutant fibrinogen, defective export and plasma deficiency. The hepatic storage predisposes to the development of liver disease. This condition has been termed hereditary hypofibrinogenemia with hepatic storage (HHHS). So far, seven of such mutations in the fibrinogen γ chain have been detected. We are reporting on an additional mutation occurring in a 3.5-year-old Turkish child undergoing a needle liver biopsy because of the concomitance of transaminase elevation of unknown origin and low plasma fibrinogen level. The liver biopsy showed an intra-hepatocytic storage of fibrinogen. The molecular analysis of the three fibrinogen genes revealed a mutation (Fibrinogen Trabzon Thr371Ile) at exon 9 of the γ chain in the child and his father, while the mother and the brother were normal. Fibrinogen Trabzon represents a new fibrinogen γ chain mutation fulfilling the criteria for HHHS. Its occurrence in a Turkish child confirms that HHHS can present in early childhood and provides relevant epidemiological information on the worldwide distribution of the fibrinogen γ chain mutations causing this disease. By analyzing fibrinogen crystal structures and calculating the folding free energy change ($\Delta\Delta G$) to infer how the variants can affect the conformation and function, we propose a mechanism for the intracellular aggregation of Fibrinogen Trabzon and other γ -module mutations causing HHHS.

Keywords: fibrinogen storage disease; Fibrinogen Trabzon; molecular modelling; folding free energy change; genetics

Fibrinogen Structural Variants Volume 3

Michael Brown



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